



### **In Memoriam**

**Dr. Petrus Johannes Waardenburg**

**3 June 1886 – 23 September 1979**

Dr. Waardenburg, the world renowned ophthalmologist and geneticist, died on 23 September 1979 in his 94th year.

Dr. Waardenburg studied medicine at the University of Utrecht and specialized in ophthalmology at the first Clinic for Eye Diseases founded in Holland by Donders. His thesis, "Investigations on the Heredity of the Physiological and Pathological Characteristics of the Eye," demonstrated early in his career his interest in genetic problems, which, throughout his long life, remained his constant interest and true passion. In 1913, he established himself as ophthalmologist in Arnhem, also practising in the country districts, and began to collect systematically observations of genetic interest, which over the years reached considerable proportions.

In 1934, he was appointed Lecturer in Human Genetics, a title he held until the age of 66 (1952). Only then was he officially appointed Professor of Genetics at the Institute of Preventive Medicine in Leyden. Nothing better illustrates the lack of consideration and esteem in which genetics was held by the universities and the physicians of his country and also by other European universities.

The year 1940 was marked by the German occupation of his country. During this period Dr. Waardenburg wrote several articles against Hitler's racist policy and anti-Semitism, illustrating his courageous and objective character.

His life was divided between his ophthalmological consultations and his scientific work on the one hand, and his devotion to his large family on the other. His modesty and courtesy hid a strong personality and an immense capacity for work and the gift of recognizing the slightest pathological variation in the eye and its adnexa.

In 1932, his book *Das Auge und seine Erbanlagen (The Eye and Its Hereditary Basis)* was published, demonstrating his encyclopaedic knowledge in the field of ophthalmology, craniofacial anomalies, and the study of twins. What is particularly striking, however, is the fact that at that time he advanced the theory of a chromosomal aberration in Mongolism, even suggesting a nondisjunction or translocation as cause of the malformation. It was only 27 years later that this theory was confirmed by Lejeune and co-workers.

From among more than 250 papers published after this monograph, I refer only to his studies on refraction, myopia, Leber's optic atrophy, and his contributions to the study of the different types of albinism. He devised a screening method for latent carriers (heterozygotes) of universal albinism, by scleral transillumination of the eye, which other authors confirmed. He was also interested in the nosology of different types of craniofacial dysostoses and contributed to this field with the description of a new type.

Throughout his entire career he dealt extensively with the study of twins and concordance and discordance rates of monozygotic and dizygotic twins for a series of hereditary diseases of the eye.

However, what really made him famous in the medical world was his description of a syndrome consisting of lateral displacement of the inner canthi and lacrimal puncta, a white forelock, a prominent, broad nasal root, heterochromia irides, and congenital deafness.

Thanks to studies in the same field and at the same time by Professor Franceschetti and myself in Geneva, scientific and personal relations were established between Dr. Waardenburg and the Geneva Eye Clinic, resulting in the joint publication of *Genetics in Ophthalmology* in two volumes in 1961 and 1963.

An indefatigable worker, Dr. Waardenburg still found time to devote himself to philosophical, religious, archeological, and political problems. He received many honours; among others, he became honorary member of several international scientific societies; he was awarded the Snellen medal, and was elected *Doctor honoris causa* of the Universities of Leyden and Munster.

Dr. Waardenburg will be remembered as a man of absolute probity, with a highly original mind, and will join the ranks of pioneers of genetic ophthalmology along with Nettleship, Julia Bell, Franceschetti, and Krill. We are saddened by his passing and express our sincere condolences to Mrs. Waardenburg and her family

David Klein  
University of Geneva